21. (Three times amended) A kit for the *in vitro* detection of a truncation, a deletion or a mutation in the survival motor neuron gene, comprising:

a set of primers wherein said primers are contained within the sequence of nucleotides 921 to 1469 of SEQ ID NO: 12 and are suitable for amplification of a fragment of said sequence;

reagents for amplifying DNA with said primers; and

a probe for the detection of the amplified product.

30. (Three times amended) A method for detecting the presence or absence of a truncation, a deletion or a mutation in the Survival Motor Neuron gene in a DNA sample, said method comprising:

(a) amplifying said DNA in the sample with primers, wherein said primers are contained in the sequence of nucleotides 921 to 1469 of SEQ ID NO: 12 and are suitable for amplification of a fragment of said sequence;

(b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP) analysis, wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample to detect alterations in the patient gene; and

(c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene.

33. (Twice amended) A method for detecting the presence or absence of Spinal Muscular Atrophy in an individual, said method comprising :

(a) hybridizing a DNA sample obtained from the individual with a DNA probe comprising all or part of the DNA sequence of SEQ ID NOS: 12 or 13 under conditions having the stringency of 10% Dextran Sulphate Sodium, 1M NaCl, 0.05M Tris-HCl pH 7.5, 0.005M EDTA and 1% SDS at 65°C ;

I3 CONT.

(b) detecting the hybrids formed, wherein the absence of detectable hybrids is indicative of the presence of Spinal Muscular Atrophy in the individual.

I4

43. (Twice amended) The method of claim 40, wherein all or part of the T-BCD541 gene is subjected to PCR amplification prior to analyzing the gene for alterations in exon 7 or 8.

I5

47. (Once amended) The method of claim 40, wherein said analyzing comprises subjecting said patient T-BCD541 gene to restriction cleavage with *Bsr*I, *Hind*III, *Sac*I or *Kpn*I.

48. (Once amended) The method of claim 40, wherein said analyzing comprises subjecting said patient T-BCD541 gene present in said biological sample to single strand conformation polymorphism analysis, wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample to detect alterations in the patient gene.

Sub K3

I6

53. (Twice amended) A kit for the *in vitro* detection of a truncation, a deletion or a mutation in the Survival Motor Neuron gene, wherein said kit comprises a probe which comprises at least 9 nucleotides within a sequence of SEQ ID NO: 12 or 13 or hybridizes with a sequence of SEQ ID NOS: 1, 2, or 10-13 under conditions having the stringency of 10% Dextran Sulphate Sodium, 1M NaCl, 0.05M Tris-HCl pH 7.5, 0.005M EDTA and 1% SDS at 65°C.

54. (Twice amended) A method of identifying the presence or absence of a mutation in the Survival Motor Neuron (SMN) gene in a nucleic acid sample, comprising

I6 CONT'D

(a) subjecting the nucleic acid in the sample to digestion by a restriction endonuclease, wherein restriction fragments resulting from said digestion of a mutated SMN gene differ from those obtained from a T-BCD541 gene of SEQ ID NO:22; and

(b) identifying the presence or absence of a mutation in the SMN gene in the subject.

I7

58. (Twice amended) A method of identifying the presence of Spinal Muscular Atrophy (SMA) in a subject, said method comprising:

(a) identifying a mutation in a T-BCD541 gene (SEQ ID NO:22) in a DNA sample obtained from said subject;

wherein the presence of a mutation in the T-BCD541 gene is indicative of the presence of SMA in said subject.

Amd K15

I8

64. (Twice amended) A kit for the *in vitro* detection of a defect in the Survival Motor Neuron gene, comprising:

a set of primers wherein said primers comprise a sequence selected from SEQ ID NOS: 5 to 8 and 24 to 57;

PCR reagents for amplifying DNA with said primers; and

a probe for the detection of the amplified product.

65. (Twice amended) A method for detecting the presence or absence of a truncation, a deletion or a mutation in the Survival Motor Neuron gene, wherein the gene is present in a DNA sample obtained from an individual, said method comprising :

(a) amplifying said DNA with primers, wherein said primers are selected from the group of SEQ ID NOS: 5 to 8;

(b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP) analysis, wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient DNA sample to a pattern of DNA fragments obtained from a control DNA sample; and

I8 cont (c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene.

Please add the following new claims 66-69.

I9 - - 66. (New) A method for detecting the presence or absence of a truncation, a deletion or a mutation in the Survival Motor Neuron gene, wherein the gene is present in a DNA sample obtained from an individual, said method comprising :

(a) amplifying said DNA with primers, wherein said primers are selected from the group of SEQ ID NOS: 24 to 57;

K16 (b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP), wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample; and

(c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene. - -

- - 67. (New) A method for detecting the presence or absence of a truncation, a deletion or a mutation in the Survival Motor Neuron gene, wherein the gene is present in a DNA sample obtained from an individual, said method comprising :

(a) amplifying said DNA with primers, wherein said primers are selected from the group of sequences which are inverted complementary sequences to SEQ ID NOS: 5 to 8;

(b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP), wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample; and

(c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene. - -

- - 68. (New) A method for detecting the presence or absence of Spinal Muscular Atrophy in an individual, comprising the steps of:

(a) contacting a biological test sample obtained from the individual with a nucleic acid probe comprising all or part of SEQ ID NO: 12 or 13, or a complement of SEQ ID NO: 12 or 13, wherein the nucleic acid probe detects a truncation, deletion or mutation of SEQ ID NO: 12 or 13,

I9 CONT.

(b) maintaining the test sample and the nucleic acid probe under conditions suitable for hybridization;

(c) detecting hybridization between the test sample and probe; and

(d) comparing hybridization in the test sample to a control sample, wherein no detectable hybridization between the test sample and probe is indicative of the presence of Spinal Muscular Atrophy in the individual. - -

- - 69. (New) A method for detecting the presence or absence of Spinal Muscular Atrophy in an individual, comprising analyzing a DNA sample obtained from the individual, wherein the DNA sample comprises the Survival Motor Neuron gene and wherein the method comprises detecting the presence or absence of either exon 7 or exon 8, or both exon 7 and exon 8 of the gene, wherein exon 7 comprises nucleotides 340 to 401 of SEQ ID NO: 13, and exon 8 comprises nucleotides 846 to 1408 of SEQ NO: 13, wherein the absence of either or both exon 7 or 8 is indicative of the presence of Spinal Muscular Atrophy in the individual. - -